

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 2. Polymorphisms Identified in *SEPN1*

Exonic Variant	Amino Acid Change	Exon
c.425A>G	Y142C	4
c.583G>A	A195T	5
c.1173T>C	P391P	9
c.1329A>G	E443E	10
c.1506A>C	K502N	12